Neuromuscular diagnoses in a nutshell

Limb-girdle muscular dystrophy (LGMD) is a collective term for a group of rare inherited neuromuscular disorders characterized by progressive muscle wasting.

The disease primarily affects proximal muscles around the shoulders and hips.

LGMD is divided into subtypes, with recessive forms designated R1–R29 and dominant forms D1–D5.

See the overview of former and current nomenclature on page 3-5 here:

Link: "New names for limb girdle muscular dystrophies"

Limb-girdle muscular dystrophy

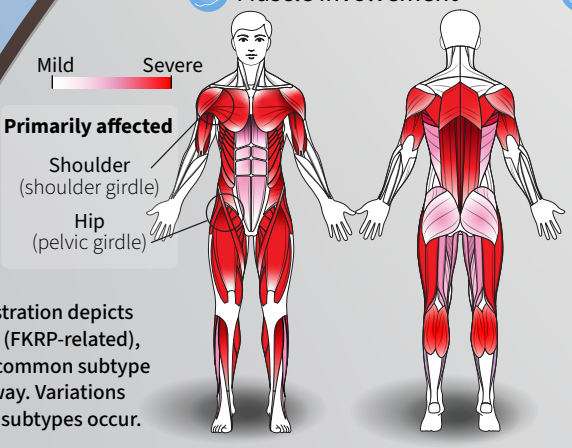
Limb-girdle muskeldystrofi

Synonym: LGMD

Classification codes: ICD-10: G71.0 | ICPC-2: N99 | ORPHA: 263*

*Each subtype has its own ORPHA code.

Muscle involvement



The illustration depicts LGMD R9 (FKRP-related), the most common subtype in Norway. Variations between subtypes occur.

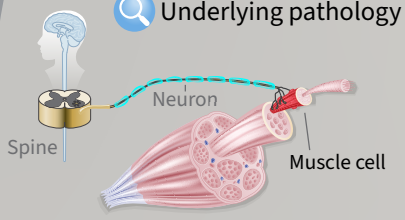
Other possible features

Cardiac involvement	Subtype dependent
Respiratory insufficiency	Subtype dependent
Scoliosis	Especially in LGMD R1
Distal involvement	Reported in LGMD R2/R12
Fatigue	

Typical initial symptom

The first symptom is usually weakness of the pelvic girdle muscles.

Underlying pathology



Caused by inherited genetic variants affecting muscle cells.

This results in progressive muscle atrophy and replacement of muscle tissue by fat and connective tissue.

Onset

Newborn ✗ | Childhood ✓ | Adolescence ✓ | Adulthood ✓ | Old age ✓

Inheritance

Autosomal dominant	Autosomal recessive	X-linked
< 10%	>90%	0%

Varies between subtypes

Sex distribution

♀ = ♂ Men and women are affected equally

Prevalence

Worldwide: approximately **1:20,000**

Treatments

*Clinical trials are ongoing

Curative treatment ✗	Gene therapy targeting the underlying cause* ✗	Medication that may slow disease progression* ✗
Physiotherapy / occupational therapy for optimization of function ✓	Assistive devices and orthopaedic aids (e.g. wheelchairs and orthoses) ✓	

Important clinical considerations

Particular caution should be exercised in patients with cardiac or respiratory involvement, and during anaesthesia.

More diagnostics information

NORWEGIAN CENTRE FOR RARE DISEASES

Sources:
- Norwegian Centre for Rare Diseases
- Orphanet
- GeneReviews®
- Socialstyrelsen
- ERN EURO-NMD
- Norwegian El. Medical Handbook (NEL)
- Treat NMD

Updated January 2026